TWO CASES OF MUSCULAR DYSTROPHY WITH NECROPSY.

(One of the Facio-Scapulo-Humeral Type Reported Clinically by Duchenne, of Boulogne ; Landouzy and Dejerine).[1]

By William G. Spiller, M.D.

Abstract.

The first case was published by Duchenne, of Boulogne, in 1872, when the patient was nine years old, and later by Landouzy and Dejerine—also as a clinical observation—when he was twenty-eight years of age. The man died in the service of Dr. Dejerine at the Bicêtre Hospital, in Paris, about six years after the second clinical observation was published. The pathological material was given by Dr. Dejerine to Dr. Spiller for microscopical study.

The case was one of hereditary progressive muscular atrophy, begining in the face when the patient was three years old. The atrophy was most pronounced in the face, shoulders and upper limbs. Permanent contractures existed in the extremities, but there were no pains, no fibrillary contractions, no hypertrophy, and no reaction of degeneration. At the necropsy the muscles were examined individually by Dr. Dejerine, and the atrophy was found to be intense. While many of the muscles of the face were greatly affected, the muscles of mastication and of the eyeballs were remarkably well preserved. The examination of all the muscles in detail will be published later. The intramuscular branches of the facial nerve, examined in the fresh state by Dr. Dejerine, and stained with osmic acid, appeared to be normal. The anterior and posterior roots, the nerves of certain muscles of the extremities, examined in the same way, were found to be normal. The nervous system studied by Dr. Spiller was found to be normal. Notwithstanding the intense atrophy of the muscles supplied by the seventh nerve, *the intramedullary portion of the facial nerve was perfectly normal on each side. The cells of the facial nucleus were large, had well-defined processes and centrally situated nuclei.*

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[1] Read at the twenty-fifth annual meeting of the American Neurological Association, June 14 and 15, 1899.

This paper will form a part of the volume soon to be issued from the William Pepper Laboratory of Clinical Medicine.

The diaphragm was found greatly degenerated, but its intramuscular nerve bundles were normal. Although the deltoid muscle was merely a mass of connective tissue, the muscle spindles and intramuscular nerve fibers appeared to be normal.

The second case was that of a man who was in the clinic of Dr. Dejerine, at the Bicêtre. In this case the face was intact. The paralysis began quite acutely in the lower limbs, as the pseudohypertrophic form, when the boy was eight years old, and extended to the trunk and upper limbs, and the patient became almost completely paralyzed in the trunk and extremities. He died at the age of twenty. The muscles were found to be intensely atrophied, but the central and peripheral nervous systems were normal. In some sections of the spinal cord a few motor cells, which might be regarded as somewhat atrophied, were seen in the anterior horns.

Dr. Spiller gave a brief review of the most important literature on the Landouzy-Dejerine form of muscular dystrophy. He spoke especially of the atrophy of the diaphragm, showing that muscles believed at one time to be spared in the atrophic process may be involved; he spoke also of the presence of normal nerve fibers and normal muscle spindles within intensely degenerated muscular tissue, as showing that the disease is probably primarily muscular. He showed that muscular atrophy does not essentially differ in the various morbid processes, and that no histological findings can be regarded as absolutely pathognomonic of any form of muscular atrophy. He spoke of the great difficulty in some cases of making a clinical diagnosis in regard to the form of atrophy, and referred to recent papers in which this difficulty is so clearly illustrated. He called attention to the bulbar form of muscular dystrophy (Hoffmann) and to the infantile family form of bulbar paralysis of Fazio and Londe, in relation to the Landouzy-Dejerine type of muscular dystrophy. The cases of congenital facial paralysis reported by H. M. Thomas were also of difficult interpretation.

Dr. Spiller then discussed the subject of muscular dystrophy from a pathological standpoint, and referred to the cases in which the nervous system was found diseased. He gave as his conclusions:

1. Cases of muscular atrophy occur which present the well-known localizations regarded as characteristic of the different forms of muscular dystrophy. In the large majority of these cases the spinal cord and peripheral nerves are normal.

2. In other rare cases presenting the clinical features of muscular dystrophy, the nervous system is more or less involved.

3. The histologic changes in the muscles are not pathognomonic of any form of atrophy.

4. It is proper to regard muscular dystrophy as a disease usually distinct from spinal muscular atrophy, but transitional forms connect the myopathic and myelopathic types of atrophy.

DISCUSSION.

Dr. H. M. Thomas said in regard to the cases he reported last year, that when he first saw them the thought of their being juvenile muscular dystrophy occurred to him at once, and the reason he did not so consider them was, in the first place, that the histories as definitely as they could be gotten showed the condition to be congenital. It was noticed directly after birth. Then the distribution of the affected muscles was exactly the same as in the cases of congenital unilateral facial paralysis combined with affection of the ocular muscles. Then too, electrical examination of the affected muscles gave no response to any amount of current, while the other muscles in the facial group gave the normal response. These symptoms, together with dulness of hearing, which seemed to be congenital, and the fact that each had congenital abnormalities of the external ear, seemed to make the diagnosis pretty clear. They may later develop muscular dystrophy, but Dr. Thomas did not believe they would. He did not see why they should be thrown out because they were bilateral when they correspond in other respects to the congenital cases. Dr. Thomas said that the whole subject of progressive muscular dystrophy is, as Dr. Spiller had shown, extremely confusing, and his conclusions expressed the belief that Dr. Thomas had come to from reviewing the literature. There have been a good many articles written upon this subject, trying to bring together all the cases of muscular dystrophy and atrophy, but Dr. Thomas believed we shall have to wait for further investigations before we can do anything more than Dr. Spiller had done in his conclusions.

Dr. Thomas did not mean to say in his paper that in any case of congenital facial paralysis the muscles of the face had been examined. The cases he referred to in which a muscle examination had been made were cases of congenital eye-muscle defects.